Complete Summary

GUIDELINE TITLE

Prenatal diagnosis of fetal chromosomal abnormalities.

BIBLIOGRAPHIC SOURCE(S)

American College of Obstetricians and Gynecologists (ACOG). Prenatal diagnosis of fetal chromosomal abnormalities. Washington (DC): American College of Obstetricians and Gynecologists (ACOG); 2001 May. 12 p. (ACOG practice bulletin; no. 27). [76 references]

GUIDELINE STATUS

This is the current release of the guideline.

This guideline updates a previous version: American College of Obstetricians and Gynecologists (ACOG). Prenatal diagnosis of fetal chromosomal abnormalities. Washington (DC): American College of Obstetricians and Gynecologists (ACOG); 1996 Sep. (Educational Bulletin Number 228).

COMPLETE SUMMARY CONTENT

SCOPE

DISEASE/CONDITION(S)

Fetal chromosomal abnormalities including Down syndrome

GUIDELINE CATEGORY

Counseling
Diagnosis
Prevention

Risk Assessment
Screening

CLINICAL SPECIALTY

Medical Genetics
Obstetrics and Gynecology

INTENDED USERS

Physicians

GUIDELINE OBJECTIVE(S)

- To aid practitioners in making decisions about appropriate obstetric and gynecologic care
- To provide clinical management guidelines for the prenatal detection of the most common autosomal trisomy in liveborn infants, Down syndrome

TARGET POPULATION

Patients at risk of fetal aneuploidy including

- Women with singleton pregnancies age 35 years and older
- Women with twin pregnancies age 33 years and older
- Women with a previous pregnancy complicated by an autosomal trisomy or sex chromosome aneuploidy
- A major fetal structural defect identified by ultrasonography
- Either parent with a chromosome translocation
- Carriers of a pericentric chromosome inversion or parental aneuploidy

INTERVENTIONS AND PRACTICES CONSIDERED

1. Second-trimester screening including maternal serum screening for alpha fetoprotein (AFP), human chorionic gonadotropin (HCG), and estriol levels and ultrasound screening

   Note: Early amniocentesis (

2. Diagnostic tests including amniocentesis, chorionic villus sampling (CVS), and cordocentesis
3. Patient counseling

MAJOR OUTCOMES CONSIDERED

- Risk factors for Down syndrome
- Risks and benefits of diagnostic procedures
- Predictive value of ultrasound markers of aneuploidy
- Predictive value of diagnostic tests for detection of fetal chromosomal abnormalities

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Hand-searches of Published Literature (Primary Sources)
Hand-searches of Published Literature (Secondary Sources)
Searches of Electronic Databases

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

The MEDLINE database, the Cochrane Library, and American College of Obstetricians and Gynecologists' (ACOG's) own internal resources and documents were used to conduct a literature search to locate relevant articles published between January 1985 and April 2000. The search was restricted to articles published in the English language. Priority was given to articles reporting results of original research, although review articles and commentaries also were consulted. Abstracts of research presented at symposia and scientific conferences were not considered adequate for inclusion in this document.

Guidelines published by organizations or institutions such as the National Institutes of Health and the American College of Obstetricians and Gynecologists were reviewed, and additional studies were located by reviewing bibliographies of identified articles.

NUMBER OF SOURCE DOCUMENTS

Not stated

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Weighting According to a Rating Scheme (Scheme Given)

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Studies were reviewed and evaluated for quality according to the method outlined by the U.S. Preventive Services Task Force:

I Evidence obtained from at least one properly designed randomized controlled trial

II-1 Evidence obtained from well-designed controlled trials without randomization

II-2 Evidence obtained from well-designed cohort or case–control analytic studies, preferably from more than one center or research group

II-3 Evidence obtained from multiple time series with or without the intervention. Dramatic results in uncontrolled experiments also could be regarded as this type of evidence.

III Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees

METHODS USED TO ANALYZE THE EVIDENCE

Review of Published Meta-Analyses
Systematic Review

DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE

Not stated

METHODS USED TO FORMULATE THE RECOMMENDATIONS

Expert Consensus

DESCRIPTION OF METHODS USED TO FORMULATE THE RECOMMENDATIONS

Analysis of available evidence was given priority in formulating recommendations. When reliable research was not available, expert opinions from obstetrician-gynecologists were used. See also the "Rating Scheme for the Strength of Recommendations" field regarding Grade C recommendations.

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Based on the highest level of evidence found in the data, recommendations are provided and graded according to the following categories:

Level A — Recommendations are based on good and consistent scientific evidence.

Level B — Recommendations are based on limited or inconsistent scientific evidence.

Level C — Recommendations are based primarily on consensus and expert opinion.

COST ANALYSIS

A formal cost analysis was not performed and published cost analyses were not reviewed.

METHOD OF GUIDELINE VALIDATION

Internal Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

Practice Bulletins are validated by two internal clinical review panels composed of practicing obstetrician-gynecologists generalists and subspecialists. The final guidelines are also reviewed and approved by the American College of Obstetricians and Gynecologists (ACOG) Executive Board.

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

The grades of evidence (I-III) and levels of recommendations (A-C) are defined at the end of "Major Recommendations" field.

The following recommendation is based on good and consistent scientific evidence (Level A):

- Early amniocentesis (

The following recommendations are based primarily on consensus and expert opinion (Level C):

- Women with singleton pregnancies who will be age 35 years or older at delivery should be offered prenatal diagnosis for fetal aneuploidy.
- Patients with a risk of fetal aneuploidy high enough to justify an invasive diagnostic procedure include women with a previous pregnancy complicated by an autosomal trisomy or sex chromosome aneuploidy, a major fetal structural defect identified by ultrasonography, either parent with a chromosome translocation, and carriers of a pericentric chromosome inversion or parental aneuploidy.
- A combination of one major or two or more minor ultrasound markers of Down syndrome substantially increases risk and warrants further counseling regarding invasive testing.
- The use of ultrasonographic screening for Down syndrome in high-risk women (e.g., women age 35 years and older) to avoid invasive testing should be limited to specialized centers.
- With an isolated choroid plexus cyst, testing is indicated only if serum screening results are abnormal or the patient will be older than 32 years at delivery.
- Cervical infections with chlamydia or herpes are contraindications to transcervical chorionic villus sampling (CVS).
- Counseling for amniocentesis in a twin pregnancy in women age 33 years is indicated because the midtrimester risk of fetal Down syndrome is approximately the same as for that of a singleton pregnancy at age 35 years.
- Nondirective counseling before genetic amniocentesis does not require a patient to commit to pregnancy termination if the result is abnormal.

Definitions:

Grades of Evidence

I Evidence obtained from at least one properly designed randomized controlled trial

II-1 Evidence obtained from well-designed controlled trials without randomization

II-2 Evidence obtained from well-designed cohort or case–control analytic studies, preferably from more than one center or research group

II-3 Evidence obtained from multiple time series with or without the intervention. Dramatic results in uncontrolled experiments also could be regarded as this type of evidence.

III Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees

Levels of Recommendation

Level A — Recommendations are based on good and consistent scientific evidence.

Level B — Recommendations are based on limited or inconsistent scientific evidence.

Level C — Recommendations are based primarily on consensus and expert opinion.

CLINICAL ALGORITHM(S)

None provided

EVIDENCE SUPPORTING THE RECOMMENDATIONS

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The type of supporting evidence is identified and graded for each recommendation (see "Major Recommendations").

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

Appropriate prenatal diagnosis of fetal chromosomal abnormalities

POTENTIAL HARMS

Adverse Effects of Diagnostic Tests:

- Amniocentesis. The fetal loss rate of traditional midtrimester amniocentesis is approximately 0.5%. Transient vaginal spotting or amniotic fluid leakage occurs in approximately 1 to 2% of all cases and chorioamnionitis occurs in less than one in 1,000 cases. Needle injuries to the fetus have been reported

but are very rare when amniocentesis is performed under continuous ultrasound guidance. Amniotic fluid cell culture is uncommon.

- Chorionic villus sampling (CVS). Risk of pregnancy loss is slightly higher with CVS than with traditional amniocentesis.

  Risk of oromandibular-limb hypogenesis is higher if CVS is performed earlier than 9 menstrual weeks.

- Cordocentesis. The pregnancy loss rate has been reported to be less than 2%.

  The fetal loss rate with amniocentesis or CVS in twin pregnancies is approximately 3.5%.

CONTRAINDICATIONS

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Some active cervical infections (such as chlamydia or herpes) are a contraindication to transcervical chorionic villus sampling (CVS).

Relative contraindications to CVS include vaginal infection, vaginal bleeding or spotting, extreme anteversion or retroversion of the uterus, and patient body habitus precluding easy access to the uterus or clear visualization of intrauterine structures with ultrasonography.

QUALIFYING STATEMENTS

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These guidelines should not be construed as dictating an exclusive course of treatment or procedure. Variations in practice may be warranted based on the needs of the individual patient, resources, and limitations unique to the institution or type of practice.

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

IOM CARE NEED

Staying Healthy

IOM DOMAIN

Effectiveness
Patient-centeredness

IDENTIFYING INFORMATION AND AVAILABILITY

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ADAPTATION

Not applicable: The guideline was not adapted from another source.

DATE RELEASED

2001 May

GUIDELINE DEVELOPER(S)

American College of Obstetricians and Gynecologists - Medical Specialty Society

SOURCE(S) OF FUNDING

American College of Obstetricians and Gynecologists (ACOG)

GUIDELINE COMMITTEE

American College of Obstetricians and Gynecologists (ACOG) Committee on Practice Bulletins-Obstetrics

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

Not stated

FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

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GUIDELINE AVAILABILITY

Electronic copies: None available

Print copies: Available for purchase from the American College of Obstetricians and Gynecologists (ACOG) Distribution Center, PO Box 4500, Kearneysville, WV 25430-4500; telephone, 800-762-2264, ext. 192; e-mail: sales@acog.org. The ACOG Bookstore is available online at the ACOG Web site.

AVAILABILITY OF COMPANION DOCUMENTS

None available

PATIENT RESOURCES

None available

NGC STATUS

This NGC summary was completed by ECRI on September 22, 2004. The information was verified by the guideline developer on December 9, 2004.

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Date Modified: 2/14/2005